

NOTES

CYANOTIC DEFECTS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Heart defects with cyanotic presentation: blue discoloration of skin/mucous membranes, typically seen at fingertips, lips, extremities
- Develop in utero
- Persistent truncus arteriosus, hypoplastic left heart syndrome, transposition of great vessels can lead to heart failure
- Persistent truncus arteriosus, tetralogy of Fallot can lead to Eisenmenger syndrome

SIGNS & SYMPTOMS

- Cyanosis
- See individual disorders

DIAGNOSIS

DIAGNOSTIC IMAGING

- Prenatal ultrasound
- Echocardiography
- Chest X-ray

OTHER DIAGNOSTICS

- ECG

TREATMENT

MEDICATIONS

- See individual disorders

SURGERY

- Definitive treatment

OTHER INTERVENTIONS

- Lifestyle changes

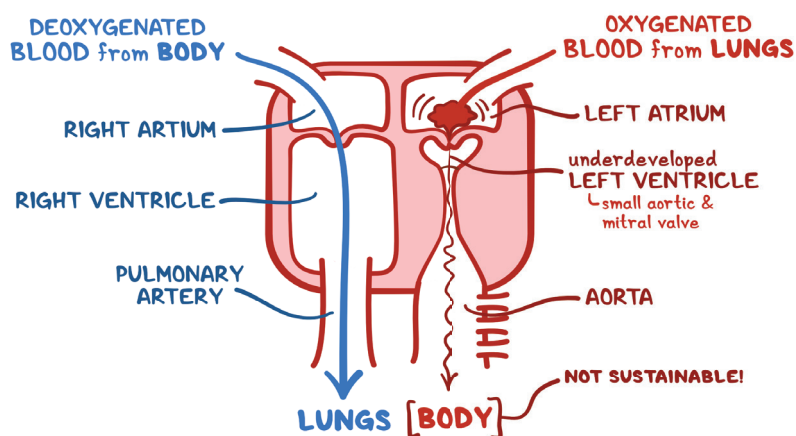


Figure 8.1 Illustration depicting blood flow in hypoplastic left heart syndrome.

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HYPOPLASTIC LEFT HEART SYNDROME

osms.it/hypoplastic-left-heart-syndrome

PATHOLOGY & CAUSES

- Congenital **underdevelopment of left heart**

CAUSES

- **Unknown:** primary congenital heart defect may reduce flow through left ventricle/ left outflow tract, affect other heart malformations
- Underdeveloped left ventricle, ascending aorta
 - Aortic/mitral valves may also be affected, narrow, or absent (atresia)
- **If untreated:** **left-sided heart failure** → cardiogenic shock → death

Atrial septal defect (ASD) and Patent ductus arteriosus (PDA)

- ASD/PDA **required** for post-natal survival in hypoplastic left heart syndrome
- **With ASD, PDA:** right heart function present but **impaired**; sometimes asymptomatic at birth
 - Oxygenated blood in left atrium flows into right atrium through ASD → pulmonary artery → PDA → aorta → body
 - **Within one day:** ductus arteriosus begins closing → **cyanosis**
- **Without ASD, PDA:** heart **not capable of sustaining life** outside womb
 - Right heart functions normally → oxygenated blood enters left atrium → flow backs up due to small mitral valve, small left ventricle → high pressure in left atrium, blood circulated ineffectively by left ventricle

SIGNS & SYMPTOMS

- Respiratory distress, poor feeding/failure to thrive, left-sided heart failure

DIAGNOSIS

DIAGNOSTIC IMAGING

Prenatal ultrasound

Chest X-ray

- Cardiomegaly

OTHER DIAGNOSTICS

ECG

- Right ventricular hypertrophy
- After birth

TREATMENT

MEDICATIONS

- **Prostaglandin E1** keeps ductus arteriosus open until surgery can be performed

SURGERY

- **Surgical repair**/heart transplant based on complexity

PERSISTENT TRUNCUS ARTERIOSUS

osms.it/truncus_arteriosus

PATHOLOGY & CAUSES

- Truncus arteriosus **fails to divide into aorta/pulmonary artery**
- Single **giant artery** branching off from right, left ventricles which splits into aorta, pulmonary artery
- Oxygenated, deoxygenated blood mix
- Deoxygenated blood mixes into systemic circulation → cyanosis

CAUSES

- Associated with **DiGeorge syndrome/22q11.2 deletion syndrome** (abnormal tissue formation during development)
- Before birth, deoxygenated blood sent to mother, oxygenated blood arrives from mother
 - Fetal heart sends blood through foramen ovale
 - Oxygenated, deoxygenated blood mix in truncus arteriosus. Both circulations get same amount of oxygenated, deoxygenated blood
 - Otherwise normal fetal development
- After birth, the baby relies on own lungs → foramen ovale closes
 - Deoxygenated, oxygenated blood still mixed → cyanosis
 - Excess **blood shunted to pulmonary circuit**, as pressure in pulmonary circuit is less than pressure of systemic circuit

RISK FACTORS

- Combination of genes, maternal environment
- Smoking, excessive alcohol intake, teratogenic medications during pregnancy; gestational diabetes; viral illness during pregnancy (e.g. German measles);

associated with genetic disorders (DiGeorge syndrome)

COMPLICATIONS

- Cardiomegaly
- Pulmonary hypertension, can progress to permanent lung damage
- Respiratory problems
- Arrhythmia
- Valve regurgitation

SIGNS & SYMPTOMS

- Difficulty breathing, pounding heart, weak pulse, poor feeding/failure to thrive, lethargy
- With physical exertion (severity varies)
 - Dizziness, fatigue, palpitations, dyspnea
- Impaired growth
- Auscultation
 - Loud systolic murmur along left sternal border due to increased flow through mitral valve
 - Constant ejection click before S2 (closure of aortic, pulmonic valves)
 - Diastolic flow murmur at apex when pulmonary blood flow increases

DIAGNOSIS

DIAGNOSTIC IMAGING

X-ray

- Shows heart size, lung abnormalities, possible presence of excess fluid in lungs

Echocardiogram

- Single large vessel arising from left, right ventricles
- Abnormalities of valves between large

vessel and ventricle it arises from

- May show abnormal blood movement between right, left ventricle, and volume of blood flow to lungs

OTHER DIAGNOSTICS

- Newborn pulse oximetry screening
 - Low oxygen saturation
 - Diagnose before symptoms develop

ECG

- Atrial enlargement (notching of P waves/P mitrale)
- Ventricular hypertrophy
- Abnormal T waves
- Right axis deviation

TREATMENT

- In rare cases, babies may survive into adulthood without surgical repair

MEDICATIONS

Diuretics

- Gets rid of excess fluid (e.g. chlorothiazide)

Inotropic agents

- Strengthens cardiac contractions (e.g. Digoxin, treats congestive heart failure, slows down heart rate, increases force of contractions)

Prophylaxis

- Antibiotics during dental/other surgical procedures to avoid infections

SURGERY

- Goal: restore normal blood flow through heart
- Procedures vary depending on individual anatomy
 - Close hole between right/left ventricles
 - Separate large vessel into pulmonary artery, aorta
 - Reconstruct single large vessel into new, complete aorta
 - Implant new tube, valve to connect right ventricle with upper part of pulmonary artery, creating new, complete pulmonary artery

OTHER INTERVENTIONS

- Lifestyle: possible limitation on intense physical activity
- Lifelong monitoring

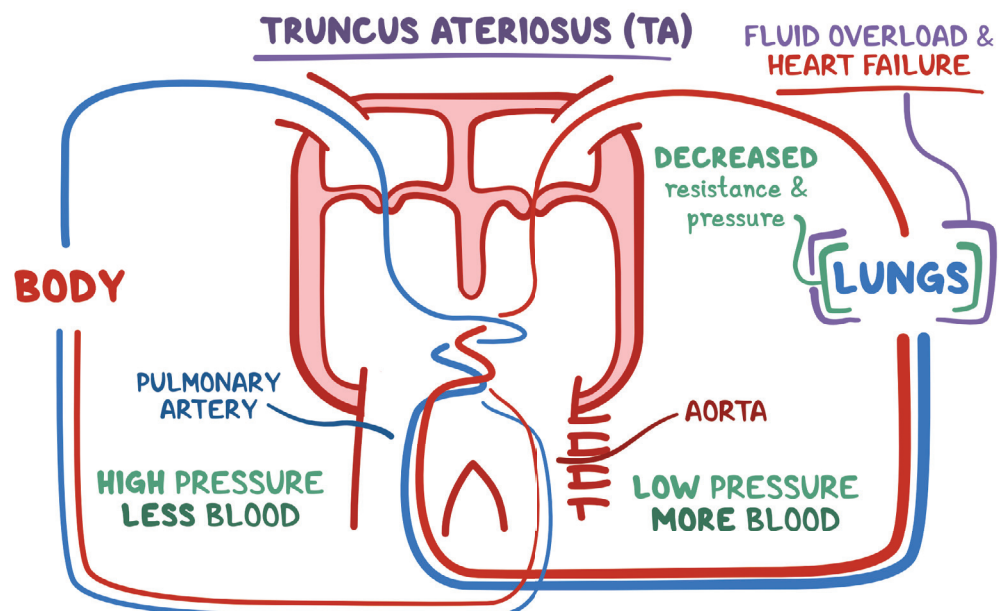


Figure 8.2 Illustration depicting blood flow through the heart in persistent truncus arteriosus.

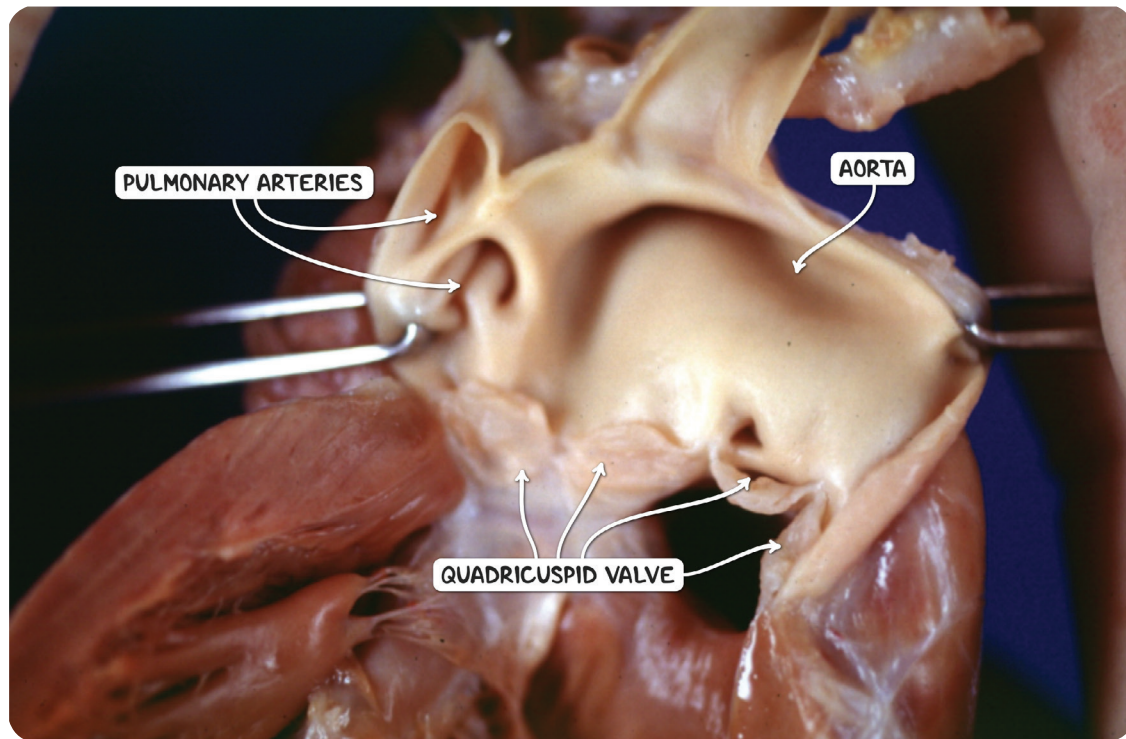


Figure 8.3 Gross pathology of a persistent truncus arteriosus. Both the left and right ventricles pump blood to both the aorta and pulmonary artery through a quadricuspid truncus valve.

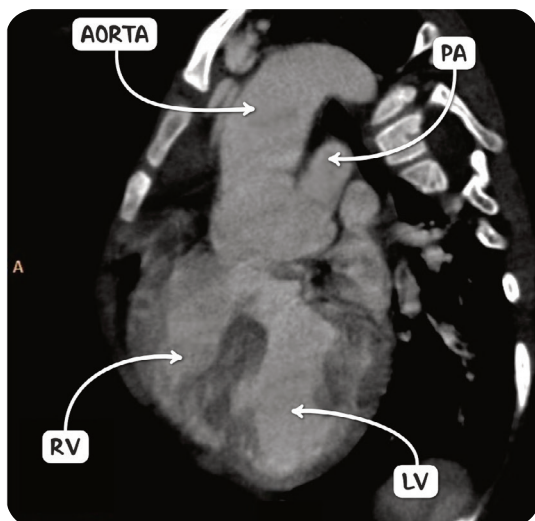


Figure 8.4 Gross pathology of a persistent truncus arteriosus. Both the left and right ventricles pump blood to both the aorta and pulmonary artery through a quadricuspid truncus valve.

TETRALOGY OF FALLOT

osms.it/tetralogy-of-fallot

PATHOLOGY & CAUSES

- Combination of **four congenital heart abnormalities**
- **Right ventricular outflow tract stenosis (pulmonic stenosis)**: obstructs pulmonary circulation
- **Right ventricular hypertrophy**: compensates for right ventricular outflow tract stenosis
- **Ventricular septal defect (VSD)**: hole in wall between ventricles. High pressure in right ventricle → blood shunts from right to left → deoxygenated blood to body
- **Aorta overrides ventricular septal defect**: aorta in abnormal position. Variable presentations

CAUSES

- Arises during cardiovascular development
- **Most common cause** of cyanotic congenital heart defects
- Four abnormalities together cause
 - **Mixing oxygenated, deoxygenated blood**
 - **Narrowed vessels/valves that increase cardiac workload**
- Severity of stenosis affects blood flow, changing pressure differentials
 - **Mild stenosis**: left-right shunt → oxygenated blood simply goes through pulmonary circulation again
 - **Severe stenosis**: right-left shunt → deoxygenated blood enters body circulation → less oxygen to tissues
- **Leads to pulmonic regurgitation**: blood flows backwards into RV, right heart overloads, can cause right-sided heart failure
- Associated with alcohol exposure in utero, maternal age 40+ years, poor nutrition or viral illness during pregnancy (e.g. rubella), Down syndrome or DiGeorge syndrome, positive family history of tetralogy of Fallot

SIGNS & SYMPTOMS

- Depend upon severity of stenosis
- Less severe right ventricular outflow obstruction often asymptomatic
- Cyanosis around lips, fingernails ("blue baby syndrome")
- Poor feeding/failure to thrive
- Harsh holosystolic murmur at left upper sternal border → sounds like pulmonary stenosis
 - Right ventricular heave
- Older infants, children
 - Clubbed fingers, toes after a few months
 - Exertional dyspnea
 - **Hypercyanotic episode (tet spell)**: on **exertion**, infant's oxygen demands increase → sudden decrease in oxygen saturation → **cyanosis**

DIAGNOSIS

DIAGNOSTIC IMAGING

Echocardiography

- Pre/postnatal

Chest X-ray

- "Boot-shaped" heart



MNEMONIC: PROVe

Components of tetralogy of Fallot

Pulmonary infundibular stenosis
Right ventricle hypertrophy
Overriding aorta
Ventricular septal defect

OTHER DIAGNOSTICS

ECG

- Right ventricular hypertrophy, right atrial enlargement

TREATMENT

MEDICATIONS

Prostaglandin E1 analogs (alprostadil)

- Severe cases
- Keep ductus arteriosus open → improve cyanosis until surgery

OTHER INTERVENTIONS

- Treat tet spell
 - *Infants squat to reduce cyanosis: kinks femoral arteries* → increases vascular resistance → increases systemic pressure → increases pressure in left ventricle to greater than pressure in right ventricle → reverse shunt to left-right → *resolve cyanosis*

SURGERY

- *Cardiac repair surgery* (first year of life)
 - VSD patch closure (only oxygenated blood flows from left ventricle into aorta)
 - Right ventricular outflow tract enlarged



Figure 8.5 Digital clubbing in an adult with tetralogy of Fallot.

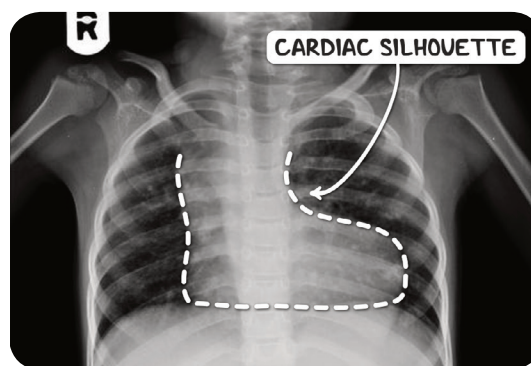


Figure 8.6 A chest radiograph of an infant demonstrating the classic boot-shaped heart seen in tetralogy of Fallot.

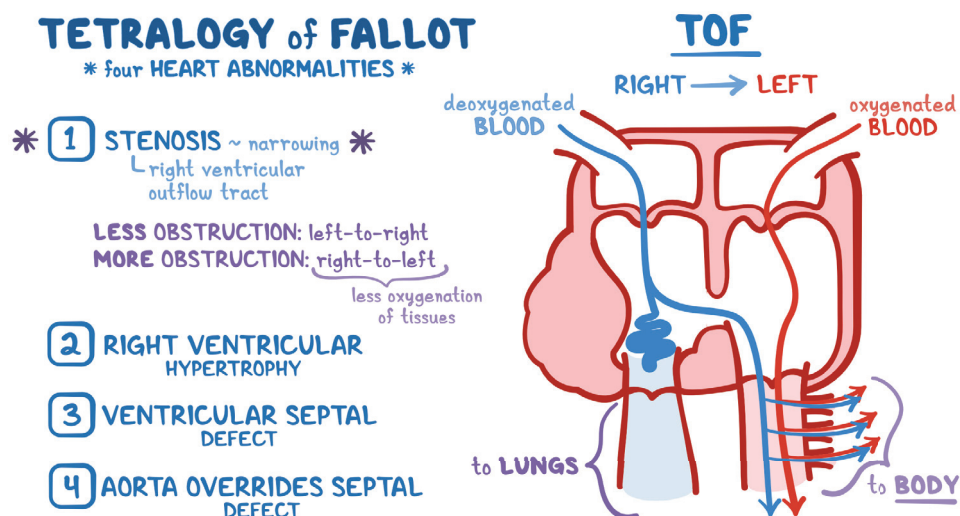


Figure 8.7 Illustration depicting blood flow through a heart with Tetralogy of Fallot.

TOTAL ANOMALOUS PULMONARY VENOUS RETURN

osms.it/anomalous-pulmonary-venous

PATHOLOGY & CAUSES

- Congenital heart defect characterized by **anomalous connection of the pulmonary veins and the heart**
- Occurs during first eight weeks of fetal development; cause unknown

TYPES

Supracardiac variant

- Most common
- Pulmonary veins open into brachiocephalic veins/superior vena cava (SVC)

Cardiac variant

- Pulmonary veins open into coronary sinus/right atrium

Infradiaphragmatic variant

- Pulmonary veins open into portal/hepatic veins

Mixed variant

- Oxygenated blood travels through **pulmonary veins to right atrium/veins** → blood does not leave pulmonary circulation → no systemic circulation
- **Incompatible with life unless foramen ovale/patent ductus arteriosus present** → oxygenated and deoxygenated blood mix → established connection between pulmonary and systemic circulation
- Anomalous connections often accompanied by **pulmonary vein obstruction** → pulmonary venous hypertension, severe cyanosis

COMPLICATIONS

- Recurrent pulmonary vein stenosis

SIGNS & SYMPTOMS

- Severity of symptoms depend upon presence/degree of obstruction
- Cyanosis, tachypnea, tachycardia, dyspnea, failure to thrive, recurrent respiratory infections
- *In case of infradiaphragmatic variant:* liver enlargement

DIAGNOSIS

DIAGNOSTIC IMAGING

Chest X-ray

- **Snowman sign (figure of 8):** dilated SVC, pulmonary vein, brachiocephalic artery forms head; dilated right atrium forms snowman's body

Echocardiography

- Right ventricular and pulmonary artery volume loading
- Might show left atrium with no connecting veins

LAB RESULTS

- Assess oxygenation and acid-base status: decreased values

OTHER DIAGNOSTICS

ECG

- Right ventricular hypertrophy

Auscultation

- Systolic ejection murmur
- Increased pulmonary component of S2
- Split S2
- S3 gallop

TREATMENT

SURGERY

- **Surgery** to establish blood flow from the right atrium to left atrium

- If present, pulmonary venous obstruction must be identified and treated promptly

OTHER INTERVENTIONS

- Cardiac catheterization

TRANSPOSITION OF THE GREAT VESSELS (TGA)

osms.it/transposition_of_great_vessels

PATHOLOGY & CAUSES

- Abnormal development causes **aorta to arise from right ventricle**, **pulmonary artery to arise from left ventricle**
- Transposition creates **two small circuits** of blood flow rather than one large
 - **Right side:** right ventricle → aorta → body → right atrium → right ventricle (blood never oxygenated)
 - **Left side:** left ventricle → pulmonary artery → lungs → pulmonary veins → left atrium → left ventricle (blood never deoxygenated)
- After birth → lungs used for oxygen → foramen ovale, ductus arteriosus close → **no exchange between two circuits** → cyanosis, death
- Sometimes, foramen ovale or ductus arteriosus stay open, or baby has ventricular septal defect (VSD); allows blood to circulate
- Different levels of severity of transposition of the great arteries (TGA)
 - **d-TGA:** dextro-TGA/complete TGA (dextro = aorta on right)
 - **l-TGA:** levo-TGA/congenitally corrected TGA (levo = aorta on left). Ventricles, valves switched. Great vessels in normal orientation, but connected to wrong ventricle. Normal blood flow circuits preserved

RISK FACTORS

- **During pregnancy:** diabetes, rubella, poor nutrition, consumption of alcohol, > 40 years old

SIGNS & SYMPTOMS

- **In utero:** asymptomatic
- **d-TGA:**
 - Cyanosis, unchanged with supplemental oxygen (less severe if VSD present)
 - Tachypnea
 - Acidosis
- **l-TGA:**
 - Asymptomatic

DIAGNOSIS

DIAGNOSTIC IMAGING

Echocardiogram

- Evaluate heart function, structure

Chest X-ray

- Classic triad
 - Heart appears as **egg on its side**/"egg on a string" appearance
 - **Lung congestion**
 - **Cardiomegaly**

Angiogram

- Pre-surgery

TREATMENT

MEDICATIONS

- Prostaglandin E: short-term solution. Keeps ductus arteriosus open

SURGERY

- Balloon atrial septostomy: short-term solution. Hole created in atrial septum
- Surgically switch great vessels
 - Five year survival rate > 80%
 - No treatment: one year survival rate 10%

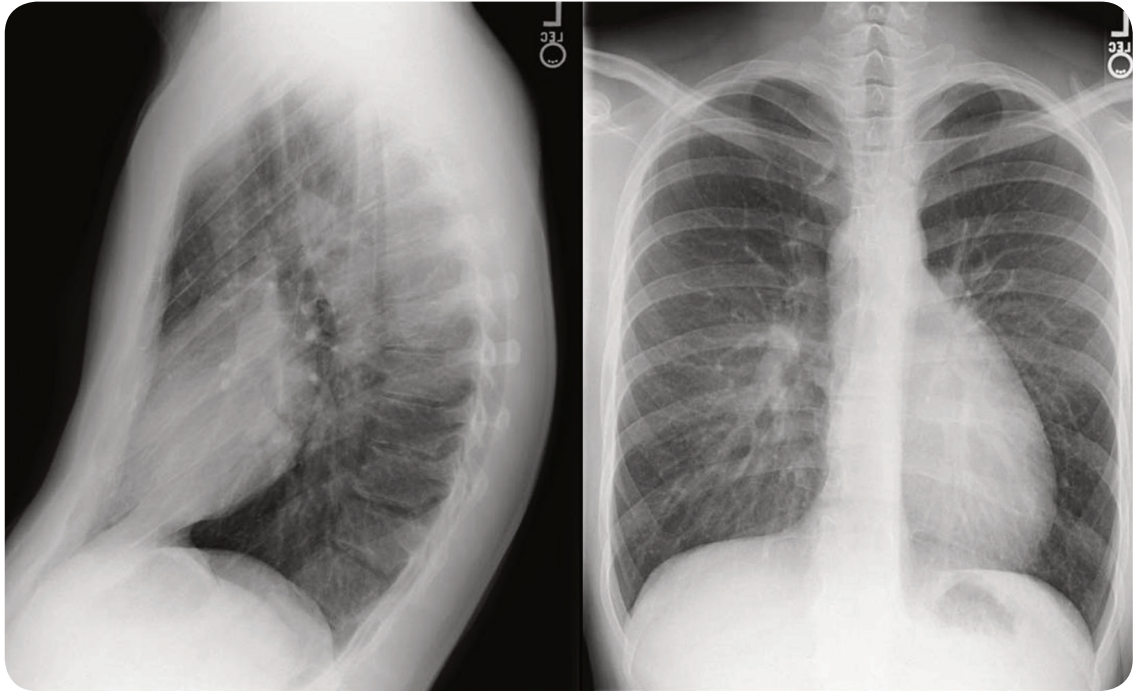


Figure 8.8 Chest radiograph in both a lateral (L) and frontal (R) view, demonstrating the “egg on a string” sign of transposition of the great vessels.

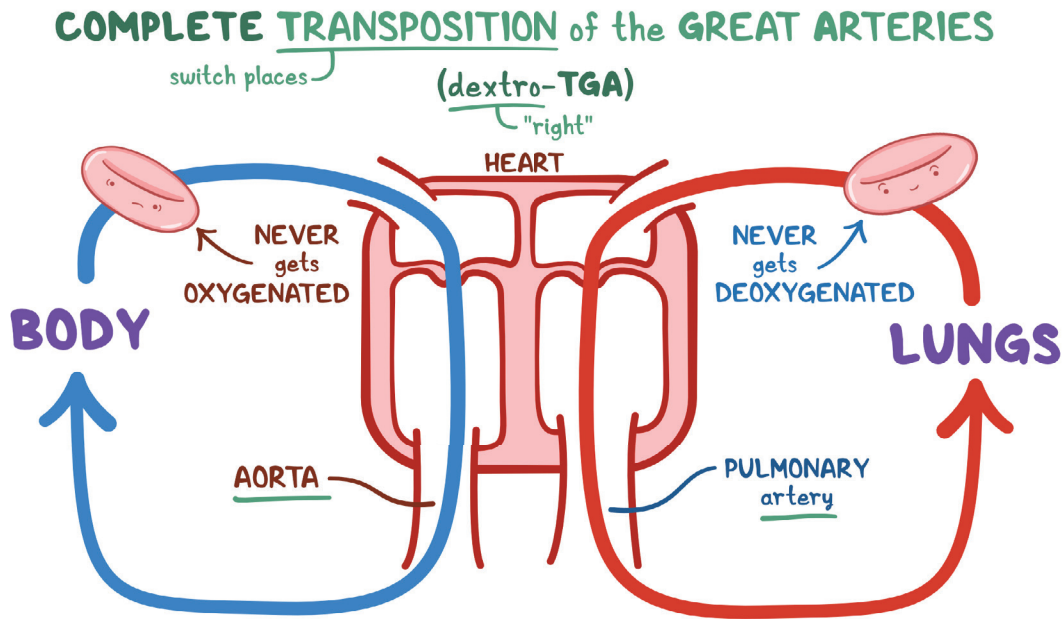


Figure 8.9 Illustration depicting blood flow through a heart with dextro transposition of the great arteries.

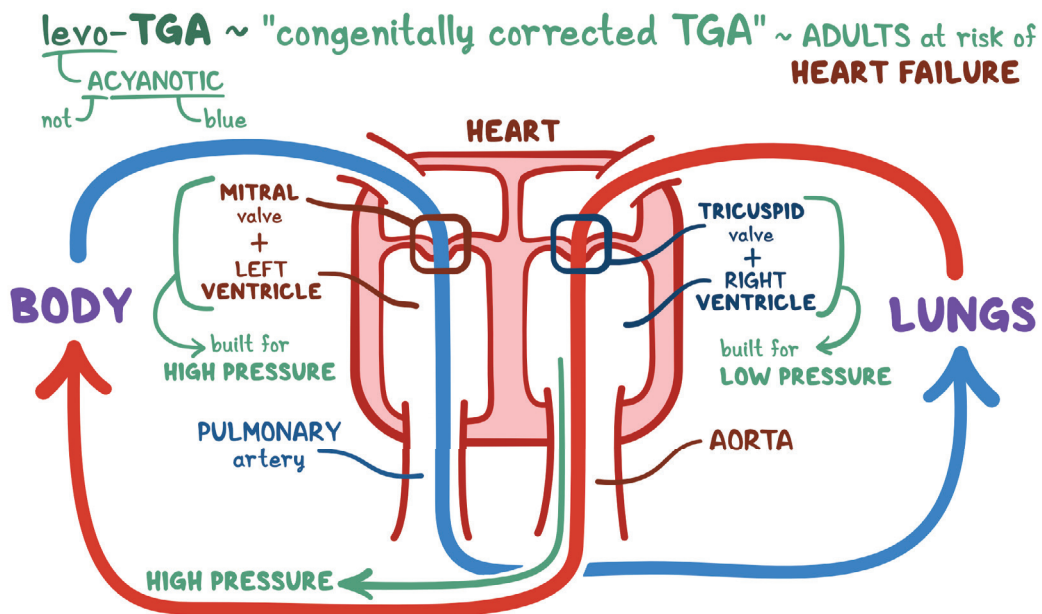


Figure 8.10 Illustration depicting blood flow through a heart with levo transposition of the great arteries.

CYANOTIC HEART DEFECT OVERVIEW

	MALFORMATION	KEY DIAGNOSTIC CLUES
HYPOPLASTIC LEFT HEART SYNDROME	Underdeveloped left side of the heart; associated with ASD and PDA for survival	CXR: Cardiomegaly ECG: Right ventricular hypertrophy Prostaglandin E required until surgery
PERSISTENT TRUNCUS ARTERIOSUS	No division causes one giant artery; associated with DiGeorge syndrome	Auscultate: Systolic murmur, ejection click Pulse Oximetry: Low ECG: Atrial enlargement, ventricular hypertrophy, right axis deviation
TETRALOGY OF FALLOT	Pulmonic stenosis, right ventricular hypertrophy, overriding aorta, VSD	Auscultate: Harsh holosystolic murmur LUSB CXR: "Boot-shaped" heart Exertional cyanosis
TRANSPOSITION OF THE GREAT VESSELS	Aorta arises from right ventricle; pulmonary artery from left ventricle	Cyanosis persists with oxygen supplementation CXR: "Egg on a string" Prostaglandin E required until surgery
TOTAL ANOMALOUS PULMONARY VENOUS RETURN	Pulmonary veins do not connect properly to heart; associated with patent foramen ovale or PDA for survival	Present